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Case report

Anaesthesia management in a patient with a severe biotinidase deficiency for congenital scoliosis repair

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KEYWORDS

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Abstract A 17 year old female patient with a biotinidase enzyme deficiency, cerebral palsy, aphamis, generalized hyperreflexia and spasticity, epilepsy and mental retardation came for the severe kyphoscoliotic deformity correction. Biotinidase enzyme deficiency is an autosomal recessive disorder with incidence of 1:60,000 neonatal birth. Treatment with biotin results in a rapid biochemical and clinical improvement. This enzyme deficiency involves neurological, neuromuscular, respiratory, dermatological and immunological problems. If untreated it can lead to convulsions, coma and death. Cobb's angle that measures the curvature of scoliosis, determined by measurements made on X rays in this case was 120° with clinical presentation of recurrent respiratory tract infection, inability to maintain sagittal posture, inability to eat or feed and difficulty in nursing care. Anaesthetic management in these patients should focus primarily on associated comorbidities and congenital anomalies affecting the course of the perioperative management and thereafter comprehensive preoperative strategies must be executed to enhance the safety profile during the surgery.

Key messages: Biotinidase deficiency is a rare congenital disorder involving multisystem. This particular case has severe scoliotic deformity, spastic cerebral palsy, mental retardation, aphamia and epilepsy. Difficult nursing care, inability to maintain sagittal posture and recurrent aspiration along with deteriorating respiratory function were the major indications for the surgical repair. A well balanced anaesthesia taking care of comorbidities and preventing trigger for malignant hyperthermia was a key for successful outcome.

1. Introduction

Scoliosis is a complex deformity of a spine involving malrotation of vertebraelateralization of a spine mainly affecting cardiovascular, respiratory and nervous systems. Anaesthetic management in this case was of special concerns due to associated comorbidities. We present this special case involving multisystem congenital disorder of Biotinidase deficiency, cerebral palsy, mental retardation, aphemia, epilepsy, severe kyphoscoliosis and its anaesthetic management. Biotinidase deficiency is a rare congenital autosomal recessive disorder with incidence of 1:60,000. Only few cases survive more than 2 years if not treated. Anaesthetic management in this case

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was a real challenge due to possible occurrence of malignant hyperthermia because of involvement of a neuromuscular system.

2. Case history

A 17 year old female patient weighing 48 kg came to the orthopaedic clinic. Patient being aphamic, parents gave the history of difficulty in sitting, difficulty in feeding, nursing care and recurrent admissions for respiratory tract infection. Patient is a known case of severe biotinidase deficiency and diagnosed at the age of 3 months due to recurrent skin lesions involving crusting, scaling and alopecia. She was started on biotin supplements and she is still on Multivitamin containing biotin 100 mg per day. The patient is also a known case of cerebral palsy, mental retardation, aphemia and epilepsy. She was previously on antiepileptic medications but currently she is not taking any medications due to no episode of seizure since 7 years.

On physical examination she has a severe thoracolumbar kyphoscoliotic deformity (Cobb's angle was 120°). Generalized hyperreflexia with spasticity was noted. Both cardiovascular and respiratory systems were normal on auscultation. She had a difficulty in lying down as well as sitting. No finding involves biotinidase deficiency such as skin lesions or alopecia. Patient cannot talk but makes incomprehensible sounds and she is mentally challenged. Power in both lower limbs was 3/5 and in both upper limbs was 5/5.

Echocardiography was advised in the view of associated congenital cardiac anomaly. Left ventricular ejection fraction was 60% with no other associated abnormal findings. Pulmonary function tests were not done as patient was mentally retarded, unable to follow commands. Other laboratory findings including renal, liver function tests and coagulation profile were normal. Serum lactate levels were normal. Urine routine examination was done to rule out aciduria and urinary tract infection as its one of the signs of biotin deficiency, found out to be normal. No finding suggests active infection. Adequate units of blood products were typed and cross matched. Antibiotic injection vancomycin 1 g was started. No other premedication was given.

After taking written informed consent of parents, patient was shifted to operation theatre. All basic monitors (electrocardiography, noninvasive blood pressure, pulse oximetry) were attached. Two broad gauze IV lines no. 16 and 18 were taken on both upper limbs. Patient was premedicated with injection midazolam 1 mg. Induction was done with injection ramifentanyl 40 μgm, injection propofol 100 mg and injection cisatracurium 4 mg. Trachea was intubated with armoured tube sized 6.5 mm. Anaesthesia was maintained with ramifentanyl and propofol target controlled infusions (TCI) pump along with air and 50% oxygen. Ventilation was volume controlled, respiratory rate was adjusted to maintain normocapnia, and muscle relaxant cisatracurium 2 mg was repeated according to train of four monitoring. Other special monitors (neuromuscular monitor, bispectral index monitor, urine output, oral temperature monitor) were attached. Left radial artery was cannulated with (no. 18) gauge arterial cannula for invasive blood pressure monitoring. Patient was placed in a prone position and all special precautions related to prone position were taken care. Temperature controlling measures such as fluid warmers and warming blankets were employed to maintain normothermia. Mean blood pressure was maintained between 55 and 60 mmHg with few injection phenylephrine 50 u boluses along with crystalloids ringer lactate boluses. Two hourly arterial blood gases were done and no signs of severe metabolic acidosis were reported. Surgery lasted for 10 h with approximate blood loss of 1.5 L which was adequately replaced with the blood by cell salvage technology. Post-operative pain was taken care by injection perfalgan 1 g IV and injection morphine 10 mg.

Patient was transferred to intensive care unit postoperatively and ventilated for next 4 h in the view of hypothermia (35°) due to prolonged surgery, which was not responding to aforementioned warming measures. Patient was sedated with fentanyl infusion 2 μg/kg/min. She was extubated uneventfully after four hours and kept in ICU for observation.

Intraoperative Blood Gases I:

Time (2 h apart)	2 h	4 h	6 h	8 h
pH	7.421	7.393	7.364	7.381
Pco2	32.4 mmHg	37.2	34.0	32.0
Po2	241.5 mmHg	248.6	254.8	265.0
HCO^3	20.6 mmol/L	22.2	18.9	18.6
BASE (effe)	−3.2 mmol/L	−2.4	−5.8	−5.7
SO2	99.8%	99.8	99.8	99.8
HB gm%	9.7 g/dl	8.1	9.1	10.1
Serum Na^+	141 mmol/L	139.2	138.5	137.3
K^+	3.12 mmol/L	3.75	3.42	3.30
Cl	103.6 mmol/L	105.5	107.6	104.5
Ca^2	1.116 mmol/L	1.097	1.082	0.912

3. Discussion

Biotinidase deficiency is an autosomal recessive inherited metabolic disorder, which was first described in 1985 by Wolf et al. The incidence of biotinidase deficiency (BD) is approximately 1:60:000 newborns in the world. Clinical manifestations of BD include neurological (seizures, ataxia, hypotonia, developmental delay, hearing and vision loss), neuromuscular (muscle weakness, spinal cord diseases), dermatological (seborrheic dermatitis, alopecia, skin rash), and metabolic abnormalities (chronic lactic acidosis, organic aciduria). It is also associated with respiratory problems (apnoea, dyspnoea, and tachypnea) and immune deficiency findings. Treatment with biotin results in a rapid clinical and biochemical improvement. However, if untreated, the disease can lead to coma and death [1]. This patient has been diagnosed at the age of 3 months and since then she is on biotin supplements. Currently she is taking multivitamin containing biotin 100 mg daily. The main anaesthesia concerns in such patients are due to metabolic acidosis, possible treatment of malignant hyperthermia due to neuromuscular system involvement, prolonged ventilation and respiratory failure [2]. Due to above concerns patient's arterial blood gases were monitored 2 hourly, and no inhalational anaesthetics were administered. Patient was induced and anaesthesia was maintained with TIVA and monitored with bispectral index which was kept in between 40 and 60 to avoid awareness [3].

Anaesthetic considerations in scoliosis surgery need special attention due to the concerns pertaining to nature of surgical intervention and coexisting multiple diseases. Severe scoliosis may itself affect nearly all physiological systems and its association with severe congenital anomalies may further increase the risk. Patient was a diagnosed case of spastic CP. The perioperative management strategies in these patients mainly focus on cardiopulmonary optimization, postoperative pain management and active measures to prevent hypothermia and seizures [4]. Most of these patients are on anticonvulsants and thus epileptogenic agents such as ketamine, pethidine, tramadol, etomidate and enflurane should be avoided. All special precautions concerned with prone position such as care of eyes, endotracheal tube, posture, neck, abdomen, nerves and pressure points were taken care of [5].

Central vascular access and arterial cannulation were difficult in this case due to contractures and improper posture. We cannulated her by two broad gauze IV lines and arterial cannulation was done in a left radial artery. Postoperative pain management needs multimodal regimen and active counselling of patients because of depressed intellectual disability or poor verbal communication skills [6]. Here the patient was mentally challenged with poor IQ and aphemia. Pain was taken care of IV morphine. Active warming measures should be initiated before induction of anaesthesia as these patients are particularly prone to hypothermia owing to extensive and prolonged surgery and their inability to regulate temperature. Besides warming measures they may require postoperative care as observed in this case who was electively ventilated for 4 h and kept in intensive care unit for observation [7].

Literature shows association of MH with myopathies in patients having scoliosis. MH is one of the clinical entities triggered by depolarizing muscle relaxant suxamethonium and nearly all volatile anaesthetic agents. We therefore adopted TIVA technique with cisatracurium small doses according to train of four monitoring for the aforementioned reasons. Blood loss is another concern in scoliosis surgery. Common strategies to minimize blood loss in perioperative period include cell saver, recombinant factor VIIa and antifibrinolytic agents (aprotinin, tranexamic acid, and epsilon aminocaproic acid). As the cell salvage technology was available, we opted for this particular technique. In cell saver 25,000 IU of heparin is used per litre of blood collected. This blood is then filtered, washed and given to the patient via blood warmers [8].

4. Conclusion

Anaesthetic management in these patients with this rare condition should focus primarily on optimization of cardio respiratory function, prevention of perioperative hypothermia and management of postoperative pain and extensive blood loss. Anaesthetic drugs should be judiciously administered keeping in mind the implications of biotinidase deficiency, epilepsy and MH. Comprehensive pre-operative strategy and perioperative management of all coexisting diseases will enable a successful outcome during the surgery.

Conflict of Interest

Author states that there is no conflict of interest.

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